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Correction: Initial experience from a renal genetics clinic demonstrates a distinct role in patient management

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The original PDF version of this Article contained an error in Table **3**. For subject number 74, the column labeled “Genetic testing” should read *CFI* p.Tyr369Ser, not *CFI* p.Tyr200Ser. This has now been corrected in both the PDF and HTML versions of the Article.



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Table 3 Patients with a known genetic disease referred to the clinic for disease management (n = 19).

Subject number	Sex/age/ ethnicity	FH	Diagnosis	Basis for diagnosis	Genetic testing	Testing lab	ACMG criteria	Reason for referral
1	F/50/EUR	Yes— multiple	Fabry disease	Low α -GAL A; positive family history	GLA p.Arg227Gln	Mount Sinai, New York, NY	LP: PM1, PM2, PP2, PP3, PP5	Renal biopsy
4–1	F/56/EUR	Yes— multiple	ADPKD	Cystic kidneys, positive family history		IHIG (Kidneyseq™), Iowa City, IA		CKD f/u
6	M/21/ EUR	Yes— multiple	Fabry disease	Low α -GAL A; positive family history	GLA p.Ser297Tyr	Mount Sinai, New York, NY	LP: PM1, PM2, PM5, PP2, PP3	CKD f/u
7	M/29/ EUR	No	Cystinosis	Fanconi syndrome, renal rickets and corneal crystals in infancy		Not done		Cysteamine Rx, manage disease
8	F/59/EUR	Yes— multiple	Fabry disease	Slit lamp, positive family history	GLA p.Trp204Ter	Mount Sinai, New York, NY	P: PV51, PM1, PM2, PP3	CKD f/u
9	M/54/ AFR	Yes— multiple	Fabry disease	Low α -GAL A; positive family history	GLA p.Trp340Ter	Mount Sinai, New York, NY	P: PV51, PM1, PM2, PP3	CKD f/u
10	M/47/ EUR	Yes— multiple	Fabry disease	Low α -GAL A; positive family history	GLA p. Ala29GlyfsTer2	Mount Sinai, New York, NY	P: PV51, PM1, PM2, PP3	CKD f/u
11	F/27/EUR	Yes— sister	Cystinosis	Bone marrow biopsy positive for cystine crystals		Not done		Cysteamine Rx
19	F/23/EUR	No	Tuberous sclerosis	Clinical criteria		Not done		Manage renal AMLs
26	F/32/EUR	No	Tuberous sclerosis + TMA in pregnancy	TSC: clinical criteria	TSC 1 c.1029 +3A>G; PLG p. Thr200Ala	CHG, Cambridge, MA; MORL (Genetic Renal Panel), Iowa City, IA	VUS: PM2, PP3, PP5; VUS: PP3	Manage tuberous sclerosis
27	M/18/ EUR	Yes— multiple	Fabry disease	Kidney biopsy	GLA p.Cys63Arg	Mount Sinai, New York, NY	LP: PM1, PM2, PM5, PP2, PP3	CKD f/u
28	M/34/ EUR	No	Fabry disease	Symptoms; positive family history	GLA p.Gly260Glu	Mount Sinai, New York, NY	LP: PM1, PM2, PM5, PP2, PP3	CKD f/u
31	M/24/ EUR	No	Unilateral renal aplasia	Antenatal and postnatal imaging		Not done		CAKUT f/u
37	M/59/ EUR	Yes— multiple	Suspected Fabry, no manifestation	Low α -GAL A; positive family history	GLA p.Ala143Thr	Mount Sinai, New York, NY	LP: PM1, PM5, PP2, PP3, PP5	Referred for renal biopsy
62	F/79/EUR	Yes— multiple	Familial hypocalciuric hypercalcaemia	Hypercalcaemia, positive family history	CaSR p.Pro55Leu	Mayo Medical Lab, Rochester, MN	LP: PM1, PM2, PP2, PP3, PP4, PP5	Post-test genetic counseling
66	F/34/EUR		aHUS			4		aHUS post-transplant f/u

Table 1 continued

Subject number	Sex/age/ ethnicity	FH	Diagnosis	Basis for diagnosis	Genetic testing	Testing lab	ACMG criteria	Reason for referral
67	M/30/ EUR	No	None	TMA, genetic screening Asymptomatic	<i>CFH</i> p. Leu1189Argfs*2 Negative for <i>NPHP1</i> variant	IIHG (Kidneyseq™), Iowa City, IA	P: PV51, PM2, PP3	Preconception-counseling, spouse with <i>NPHP1</i> deletion
74	F/40/EUR	No	aHUS	TMA, genetic screening	<i>CFI</i> p.Tyr369Ser	MORL (Genetic Renal Panel), Iowa City, IA	LP: PM1, PM2, PP3, PP5	aHUS post-transplant f/u
75	F/38/EUR	No	aHUS	TMA, genetic screening	<i>CFH</i> p.Glu625Ter	MORL (Genetic Renal Panel), Iowa City, IA	P: PV51, PM2, PP3	aHUS post-transplant f/u

Genetic screening in these patients was performed prior to referral.

ACMG American College of Medical Genetics and Genomics, *ADPKD* autosomal dominant polycystic kidney disease, *AFR* African/African American, *aHUS* atypical hemolytic uremic syndrome, *AML* angiosarcoma, *CAKUT* congenital anomalies of kidney and urinary tract, *CHG* Center for Human Genetics, *CKD* chronic kidney disease, *EUR* Caucasian, *f/u* follow up, *FH* family history, *IIHG* Iowa Institute of Human Genetics, *LP* likely pathogenic, *MORL* Molecular Otolaryngology and Renal Research Laboratories, *P* pathogenic, *TMA* thrombotic microangiopathy, *TSC* tuberous sclerosis, *VUR* vesicoureteric reflux, *VUS* variant of unknown significance, α -*GAL* A α -galactosidase A.